



USP9Y gene

ubiquitin specific peptidase 9, Y-linked

Normal Function

The *USP9Y* gene provides instructions for making a protein called ubiquitin-specific protease 9. This gene is found on the Y chromosome. People normally have 46 chromosomes in each cell. Two of the 46 chromosomes are sex chromosomes, called X and Y. Females have two X chromosomes (46,XX), and males have one X chromosome and one Y chromosome (46,XY).

Because it is located on the Y chromosome, the *USP9Y* gene is present only in males. It occurs in a region of the Y chromosome called azoospermia factor A (AZFA). Azoospermia is the absence of sperm cells. The *USP9Y* gene is believed to be involved in sperm cell development, but its specific function is not well understood.

Health Conditions Related to Genetic Changes

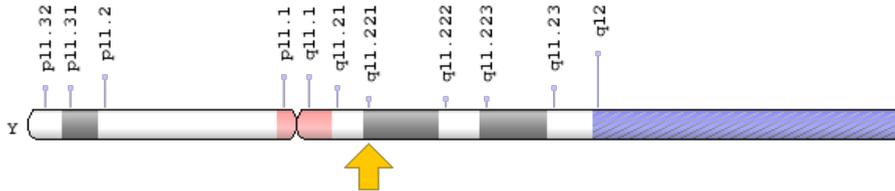
Y chromosome infertility

A small number of individuals with Y chromosome infertility have mutations in the *USP9Y* gene or deletions of all or part of the gene. These changes in the *USP9Y* gene prevent the production of ubiquitin-specific protease 9 or result in the production of an abnormally short, nonfunctional protein. The absence of functional ubiquitin-specific protease 9 impairs the production of sperm cells, resulting in an inability to father children.

Chromosomal Location

Cytogenetic Location: Yq11.221, which is the long (q) arm of the Y chromosome at position 11.221

Molecular Location: base pairs 12,701,231 to 12,860,844 on the Y chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AZF
- AZF1
- AZFA
- azoospermia factor 1
- deubiquitinating enzyme FAF-Y
- DFFRY
- fat facets protein related, Y-linked
- FLJ33043
- SP3
- ubiquitin specific peptidase 9, Y-linked (fat facets-like, Drosophila)
- ubiquitin-specific processing protease FAF-Y
- ubiquitin specific protease 9, Y chromosome (fat facets-like Drosophila)
- ubiquitin specific protease 9, Y-linked
- ubiquitin thiolesterase FAF-Y
- USP9Y_HUMAN

Additional Information & Resources

GeneReviews

- Y Chromosome Infertility
<https://www.ncbi.nlm.nih.gov/books/NBK1339>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28USP9Y%5BTIAB%5D%29+OR+%28%28AZF%5BTIAB%5D%29+OR+%28AZF1%5BTIAB%5D%29+OR+%28AZFA%5BTIAB%5D%29+OR+%28DFFRY%5BTIAB%5D%29+OR+%28SP3%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D>

OMIM

- UBIQUITIN-SPECIFIC PROTEASE 9, Y CHROMOSOME
<http://omim.org/entry/400005>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_USP9Y.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=USP9Y%5Bgene%5D>
- HGNC Gene Family: Minor histocompatibility antigens
<http://www.genenames.org/cgi-bin/genefamilies/set/870>
- HGNC Gene Family: Ubiquitin specific peptidases
<http://www.genenames.org/cgi-bin/genefamilies/set/366>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=12633
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/8287>
- UniProt
<http://www.uniprot.org/uniprot/O00507>

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